A 15-year-old Nicaraguan female diagnosed with Proteus Syndrome at 3 months old presented to a pediatric cardiologist at 5 years of age after experiencing a near-syncopal event involving pallor, dizziness, and nausea. Her medical history was only significant for multiple surgical resections of soft tissue tumors in the umbilical and in the vaginal areas. Family history was negative for any congenital or acquired cardiac disease. On examination, swelling was noted of the right pectoral area, and there was right-sided hemihypertrophy. The remainder of the examination was normal, with a heart rate of 100 and blood pressure of 100/60 mm Hg. An initial electrocardiogram revealed a low atrial rhythm with right-axis deviation, right ventricular hypertrophy with strain, and ST changes in the inferior leads. An echocardiogram showed abnormal right ventricular thickening with abnormal diastolic filling. No valvular dysfunction or flow turbulence was noted. The patient was treated with β-blockers, restricted from competitive sports and physical education, and asymptomatic from a cardiorespiratory standpoint. At 6 years of age, she developed a seizure disorder, which was well controlled with medication and ultimately discontinued. Transthoracic (Figure 1) and transesophageal echocardiography (Figures 2 and 3) demonstrated thickening, marked echogenicity, and signal inhomogeneity of the biventricular walls, and no outflow tract obstruction was observed.

A cardiac MRI was performed at 15 years of age to evaluate the progressive ventricular wall thickening seen by serial echocardiography. There was no evidence of structural heart disease or left ventricular outflow tract obstruction. The MRI was remarkable for marked biventricular and septal hypertrophy with inhomogeneity of signal intensity. Figure 4A and 4B shows midventricular 4-chamber and short-axis steady state free precession cine images, gated to diastole. In both images, there is hyperintense/bright signal of the tissue surrounding and between the left and right ventricles when compared with the normal myocardial signal intensity at the endocardial surface. Figure 5A and 5B shows T1-weighted turbo spin echo; these slices also demonstrate a markedly hyperintense signal of the same epicardial and midseptal segments. After application of a fat saturation pulse, these same segments are hypointense/dark (Figure 6A and 6B). Late gadolinium enhancement (Figure 7A and 7B) shows diffuse enhancement of this epicardial and midseptal(s) tissue with some heterogeneity in signal intensity. On the basis of the tissue characteristics demonstrated by the various sequences, we felt that this tissue was primarily fat (lipoma) with some fibrous elements. Fibrous pericardium seemed to surround the heart and lipomatous tissue fully (Figure 4A and 4B, thick arrow). Interestingly, on all of the 4-chamber images, the serous pericardium (Figures 4A–7A, thin arrow) seemed to be separating 2 planes of fat as suggested by 2 layers of bright signal intensity within the lipomatous tissue (brighter signal at the extreme epicardial surface). The coronary arteries were also seen coursing within these layers (Figure 5A, **) in the epicardial space. The tissue contrast between presumably normal myocardium and lipomatous tissue was relatively well demarcated on the steady state free precession short-axis stack, consequently contouring of the ventricular volumes and mass was attempted (Figure 8). These contours revealed normal biventricular volumes and myocardial mass (Table).

The numbers were internally consistent, in that ventricular stroke volumes were nearly identical for the left and right ventricles (56 and 55 mL/m², respectively), and the cardiac index obtained by volumetric analysis was identical to the cardiac index calculated from aortic valve flow analysis (4.0 L/min per square meter). The epicardial surface of the lipomatous tissue was also separately contoured to calculate the mass of lipoma: After subtracting the biventricular volumes and muscle volumes from the surrounding lipoma (green) contour, the remaining volume was multiplied by the specific gravity of fat (0.9) to calculate an approximate fat mass of 494 g/m², >6× the combined ventricular mass! The referring cardiologist was reassured that the apparent ventricular hypertrophy was, in fact, entirely because of lipomatous tissue and that the patient did not have any element of myocardial hypertrophic cardiomyopathy.

Proteus syndrome is a rare disorder comprised of a disproportionate overgrowth of limb, skull, and visceral tissues, lipomas, scoliosis, and joint limitation. Aortic vascular malformations have been described, but there have been few reports describing cardiac disease associated with Proteus syndrome. The first such case report was of a 20-year-old Chilean man with macrodactyly, partially amputated right index finger, cranial exostoses, protosis, severe pulmonary fibrocystic changes, sinus...
tachycardia, and right bundle branch block. His echocardiogram demonstrated thickening of the cardiac septum and an apical right ventricular mass, but no tissue characterization or histology was obtained.² The presence of fatty elements has been described by cardiac MRI in the cardiac tumor of a Japanese girl with multiple cranial hyperostoses and features suggestive of but not classic for Proteus Syndrome.³ This patient had a mass localized to the anterior right ventricular free wall with bright signal on T1-weighted images, similar to our patient.

The degree of cardiac lipomatosis, as we describe in this case, has never been reported in the literature. Cardiac MRI was critical in defining tissue characteristics but equally important in providing quantitative data that were able to define normal cardiac volumes, mass, and function, separate from the lipomatous tissue. Certainly, the clinical significance of this diagnosis is unclear because some pediatric cardiac tumors, including lipomatous masses, have been associated with ventricular arrhythmias and aborted sudden death and may result in tumor erosion through the myocardium.⁴ Most recently, this patient underwent defibrillator placement after a few episodes of syncope with no documented arrhythmias by Holter monitoring. However, an electrophysiology study was positive for inducible ventricular tachycardia.

Disclosures

None.

References


Key Words: cardiomyopathy, hypertrophic ▪ hypertrophy ▪ lipomatosis ▪ magnetic resonance imaging ▪ pediatric

Figure 1. A short-axis image of the ventricles by transthoracic echocardiogram. There is circumferential, biventricular hypertrophy, and heterogeneous echogenicity of the myocardium (•).

Figure 2. A transesophageal echocardiogram of the left ventricle and left ventricular outflow tract (LVOT), showing base to apex involvement of the left ventricular myocardium (•).

Figure 3. A transesophageal echocardiogram image, demonstrating involvement of the right ventricular free wall and conal septum as well (•). RVOT indicates right ventricular outflow tract.

Figure 4. Four-chamber (A) and short-axis (B) sequences gated to end-diastole. A and B, Steady state free precession sequences showing marked biventricular and septal hypertrophy with signal inhomogeneity; the normal myocardium is surrounded by a hyperintense signal, which is brightest at the epicardial surface. The parietal pericardium (thick arrow) completely surrounds the heart and lipomatous tissue, but there is also a layer of serous pericardium (thin arrow), which seems to separate the lipoma into 2 distinct layers with slightly different signal intensity.
Figure 5. Four-chamber (A) and short-axis (B) sequences gated to end-diastole. A and B. Turbo spin echo T1-weighted images, which reveal fatty elements as hyperintense signal surrounding normal myocardium. The right coronary artery can be seen coursing through the lipomatous tissue (**). Note that the thin arrow in A demonstrates the serous pericardium.

Figure 6. Four-chamber (A) and short-axis (B) sequences gated to end-diastole. A and B, Turbo spin echo images with application of a fat saturation pulse; the fatty tissue is suppressed and thus hypointense. Note that the thin arrow in A demonstrates the serous pericardium.

Figure 7. Four-chamber (A) and short-axis (B) sequences gated to end-diastole. A and B. Diffuse and heterogeneous gadolinium uptake in the lipomatous tissue consistent with a fibrotic component to the mass. Again note that there is septal involvement (s) best seen not only on these late gadolinium enhancement images but also throughout the 4 sequences. Note that the thin arrow in A demonstrates the serous pericardium.

Figure 8. A cine short-axis stack of the ventricles obtained in end-diastole. Endocardial contours of the left ventricle (yellow), right ventricle (red), and epicardial contours (blue) are used to calculate end-diastolic volumes and myocardial mass for each ventricle. Similar contours were performed for the ventricular endocardial borders in systole to calculate stroke volume and ejection fractions for both ventricles, as listed in the Table. A fourth contour, in green, surrounds the lipomatous tissue. The ventricular volumes and muscle volumes were subtracted from the lipoma contour volume to obtain a volume of fat. This value was multiplied by the specific gravity of fat (0.9) to obtain an estimate of lipoma mass, which was 494 g/m² when compared with a combined ventricular mass of 80.3 g/m².

Table. MRI-Derived Ventricular Volumes, Function, and Mass

<table>
<thead>
<tr>
<th></th>
<th>LV</th>
<th>RV</th>
<th>Lipomatous Tissue</th>
</tr>
</thead>
<tbody>
<tr>
<td>EF, %</td>
<td>65.1</td>
<td>66.9</td>
<td>…</td>
</tr>
<tr>
<td>EDV, mL/m²</td>
<td>86.2</td>
<td>82</td>
<td>…</td>
</tr>
<tr>
<td>ESV, mL/m²</td>
<td>30.1</td>
<td>27.1</td>
<td>…</td>
</tr>
<tr>
<td>SV, mL/m²</td>
<td>56.1</td>
<td>54.9</td>
<td>…</td>
</tr>
<tr>
<td>Mass, g/m²</td>
<td>51.4</td>
<td>28.9</td>
<td>494</td>
</tr>
<tr>
<td>Cardiac index, L/min per square meter</td>
<td>3.98</td>
<td>…</td>
<td>…</td>
</tr>
</tbody>
</table>

EDV indicates end-diastolic volume indexed; EF, ejection fraction; ESV, end-systolic volume indexed; LV, left ventricle; Mass, mass indexed; RV, right ventricle; and SV, stroke volume indexed.
Are You Calling Me Fat? An Extreme Case of Cardiac Lipomatisos Masquerading as Hypertrophic Cardiomyopathy
Eva H. Nunlist, Jorge Garcia and Ruchira Garg

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SUPPLEMENTAL MATERIAL

Video 1 is a transesophageal echocardiogram (TEE) clip of the left ventricle showing marked hypertrophy and echogenicity of the myocardium, but no significant left ventricular outflow tract obstruction.

Video 2 is a TEE clip of the right ventricle showing involvement of the right ventricular free wall and conal septum as well.

Video 3 is a steady state free precession (SSFP) cine MRI image in the 4 chamber plane. In this movie, the difference in signal from the normal myocardium (hypointense/dark) and the lipomatous tissue (hyperintense/bright) is readily apparent.

Video 4 is a SSFP cine MRI image in short axis at the mid-ventricular level showing marked concentric biventricular lipomatous tissue, including infiltration of the mid-septum. Biventricular systolic function is clearly preserved.